Langer-Giedion syndrome

Langer-Giedion syndrome is a condition that causes bone abnormalities and distinctive facial features. People with this condition have multiple noncancerous (benign) bone tumors called osteochondromas. Multiple osteochondromas may result in pain, limited range of joint movement, and pressure on nerves, blood vessels, the spinal cord, and tissues surrounding the osteochondromas. Affected individuals also have short stature and cone-shaped ends of the long bones (epiphyses). The characteristic appearance of individuals with Langer-Giedion syndrome includes sparse scalp hair, a rounded nose, a long flat area between the nose and the upper lip (philtrum), and a thin upper lip. Some people with this condition have loose skin in childhood, which typically resolves with age. Affected individuals may have some intellectual disability.

Frequency

Langer-Giedion syndrome is a rare condition; its incidence is unknown.

Genetic Changes

Langer-Giedion syndrome is caused by the deletion or mutation of at least two genes on chromosome 8. Researchers have determined that the loss of a functional *EXT1* gene is responsible for the multiple osteochondromas seen in people with Langer-Giedion syndrome. Loss of a functional *TRPS1* gene may cause the other bone and facial abnormalities. The *EXT1* gene and the *TRPS1* gene are always missing or mutated in affected individuals, but other neighboring genes may also be involved. The loss of additional genes from this region of chromosome 8 likely contributes to the varied features of this condition.

Langer-Giedion syndrome is often described as a contiguous gene deletion syndrome because it results from the loss of several neighboring genes.

Inheritance Pattern

Most cases of Langer-Giedion syndrome are not inherited, but occur as random events during the formation of reproductive cells (eggs or sperm) in a parent of an affected individual. These cases occur in people with no history of the disorder in their family. There have been very few instances in which people with Langer-Giedion syndrome have inherited the chromosomal deletion from a parent with the condition.

Langer-Giedion syndrome is considered an autosomal dominant condition because one copy of the altered chromosome 8 in each cell is sufficient to cause the disorder.

Other Names for This Condition

- Giedion-Langer Syndrome
- tricho-rhino-phalangeal syndrome type II
- trichorhinophalangeal syndrome type II
- TRPS II

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Langer-Giedion syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0023003/

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html

Genetic and Rare Diseases Information Center

- Trichorhinophalangeal syndrome type 2
  https://rarediseases.info.nih.gov/diseases/7801/trichorhinophalangeal-syndrome-type-2

Educational Resources

- MalaCards: trichorhinophalangeal syndrome, type ii
  http://www.malacards.org/card/trichorhinophalangeal_syndrome_type_ii_2
- Orphanet: Langer-Giedion syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=502
- Univeristy of Kansas Medical Center Resource List
  http://www.kumc.edu/gec/support/langer_g.html

Patient Support and Advocacy Resources

- Chromosome Disorder Outreach
  http://chromodisorder.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/trichorhinophalangeal-syndrome-type-ii/
- The MAGIC Foundation
  https://www.magicfoundation.org/
- Unique: Rare Chromosome Disorder Support Group
  http://www.rarechromo.org/html/home.asp

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Langer-Giedion+syndrome%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28Langer-Giedion+syndrome%5BTIAB%5D%29+OR+%28tricho-rhino-phalangeal+syndrome+type+II%5BTIAB%5D%29+OR+%28trichorhinophalangeal+syndrome+type+II%5BTIAB%5D%29%29+OR+%28TRPS+II%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- TRICHORHINOPHALANGEAL SYNDROME, TYPE II
  http://omim.org/entry/150230

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